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RESEARCH ARTICLE

APERT SYNDROME : A CASE REPORT

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Abstract

Apert syndrome (AS) is indeed a rare congenital disorder that falls under the category of acrocephalosyndactyly. It is characterized by several distinct features, including craniosynostosis (premature fusion of certain skull bones), midface hypoplasia (underdevelopment of the middle part of the face), and syndactyly (fusion) of the hands and feet.The syndrome is typically caused by mutations in the fibroblast growth factor receptor gene (FGFR2). These mutations are inherited in an autosomal dominant pattern, which means that an affected individual has a 50% chance of passing the syndrome on to their children.Diagnosing Apert syndrome can be challenging due to its rarity and similarities with other craniosynostosis syndromes. However, certain key features can help differentiate it from other conditions. These features include early fusion of the coronal suture , cranial base synostosis , and agenesis (absence) of the sagittal suture (the suture running along the top of the skull).The characteristic appearance and dental features of Apert syndrome include maxillary transverse and sagittal hypoplasia, dental crowding, a pseudo-cleft palate and skeletal and dental anterior open bite.Given the multiple phenotypic signs and complex nature of the syndrome, successful management of Apert syndrome requires a multidisciplinary team approach. This team typically includes professionals from various specialties such as dentistry, neurosurgery, plastic surgery, psychiatry, ophthalmology, perinatology, and genetics. Each specialist contributes their expertise to provide comprehensive care and support for individuals with Apert syndrome.

Introduction:-

Apert syndrome (AS), a form of acrocephalosyndactyly, is a rare congenital disorder that involves craniosynostosis, midface hypoplasia, and syndactyly of the hands and feet[1,2]. The syndrome was first mentioned by Baumgartner in 1842 and Wheaton in 1894, but it was French paediatrician Eugene Apert who first described a group of individuals with a similar disorder in 1906, and the syndrome was subsequently named after him.The prevalence of Apert

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syndrome is estimated to be between 1 in 65,000 and 1 in 200,000 newborns, and it does not show a predilection for a specific gender[4]. The syndrome is associated with two specific mutations in the fibroblast growth factor receptor 2 (FGFR2) gene, which is located on chromosome 10q26. The S252W mutation occurs in 67% of patients and is associated with more severe craniofacial anomalies, while the P253R mutation is linked to severe syndactyly (fusion of digits).These mutations affect the region between the immunoglobulin-like domains II and III of the FGFR2 protein, resulting in increased affinity and altered specificity of ligand binding. This leads to dysregulation of cell migration, proliferation, and differentiation, ultimately causing premature osteogenesis (bone formation) and the skeletal abnormalities characteristic of the syndrome[3,4].

Typical clinical characteristics of Apert syndrome

Craniosynostosis

The coronal synostosis and the sagittal and metopic suture agenesis accompanied with early synostosis of the cranial base results in acrocephaly, brachycephaly, flat occiput and high prominent forehead, hypoplastic midface, and a verticalcraniofacial complex.[1,2,4,7]

Midface hypoplasia.

Eyes shows downward-slanting palpebral fissures, hypertelorism, shallow orbit, proptosis, and exophthalmos. The nose has a feature that is marked flat nasal bridge. Additionally, the maxilla is hypoplastic and retro-positioned. The palate is high-arched and narrow due to poor aeration in maxillary antra.[3,5]There are bulbous lateral palatal swellings, which make the central furrow of the palate very prominent and difficult to cleanse. An anteriorly tipped palatal plane with pseudo-cleft palate along with is very common.[9,10]

Syndactyly

Syndactyly or webbing of fingers causes immobility of fingers due to ossification of interphalangeal joints due to segmentation of embryonic phalanges. Involvement of the first or fifth digits in this bony mass is common. There can be a similar deformity involving the foot called “mitten hand and sock foot”[5].

Dental Manifestation

Some of the dental characteristics associated with Apert Syndrome sequence include: Maxillary anterior open bite,Severely crowded and retrusive maxillary arch,V-shaped maxillary arch with posterior slant, Delayed eruption and impactions, Impactions, Thick gingiva, Supernumerary or congenitally missing teeth.

Case Report

An 18 year old female patient reported to Oral medicine and radiology department with a chief complaint of un-anesthetic appearance due to crowding in upper front teeth. The patient presented with not usual craniofacial and dental features, which give rise to a detailed examination of the case. This female patient has the present condition by birth. Both the parents were normal and in fourth decades of life. She was the second child from a consanguineous marriage and has one sibling who was normal and mother had a normal delivery with no history of trauma, infection, drug use during the term. Family history of similar complaints was reported in first degree paternal relative.The patient had normal developmental milestones with minimal signs of mental retardation. The speech was slightly unclear.Clinical examination reveal patient had abnormal turri-brachycephalic head contour, flat occipit,retruded maxilla. Esotropia left eye, Hypertelorism, Antimongoloid slant , cross -bow shaped lips form.Characteristics limb defect Syndactyly of hands with invandly hypoplastic thumb and syndactyly of both feet with deformation of great toe. Intraoral examination revealed Constricted v- shaped maxilla , high arch palate , anterior open bite with dental crowding .

Clinical Findings

Fig. 1:- Extraoral photograph showingStrabismus (Esotropia left eye)AndHypertelorism,Down sliding lateral palpebral fissures(AntimongoloidSlant),Cross bow-shapedlips.

Fig. 2:- Abnormal turri-brachycephalic head contour (tall and AP shortened),Flat occiput,Retruded maxilla.

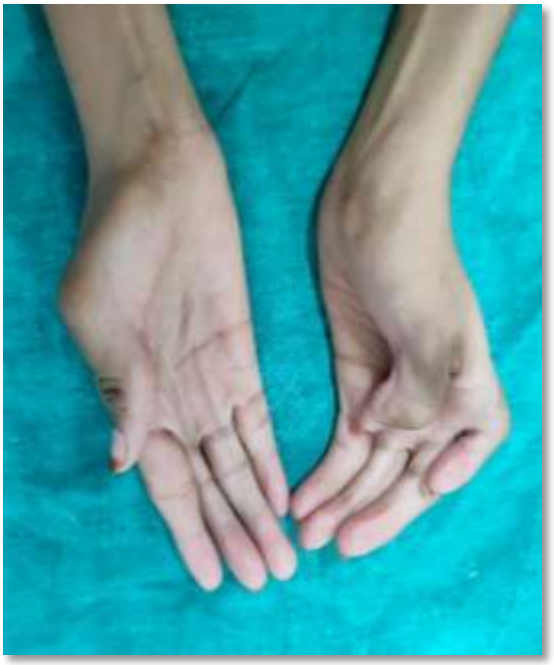


Fig3:-Syndactylyof both hands with inwardly.

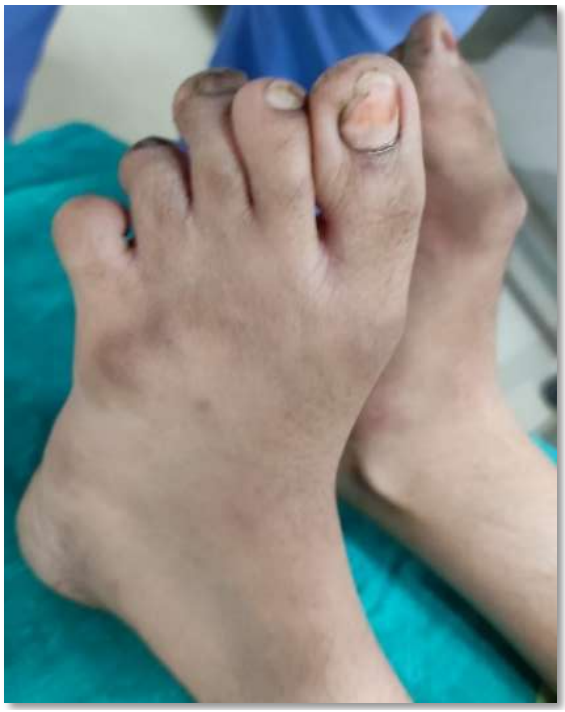


Fig 4:- Syndactyly present with both feet with deformation of the great toe.

Radiological Findings

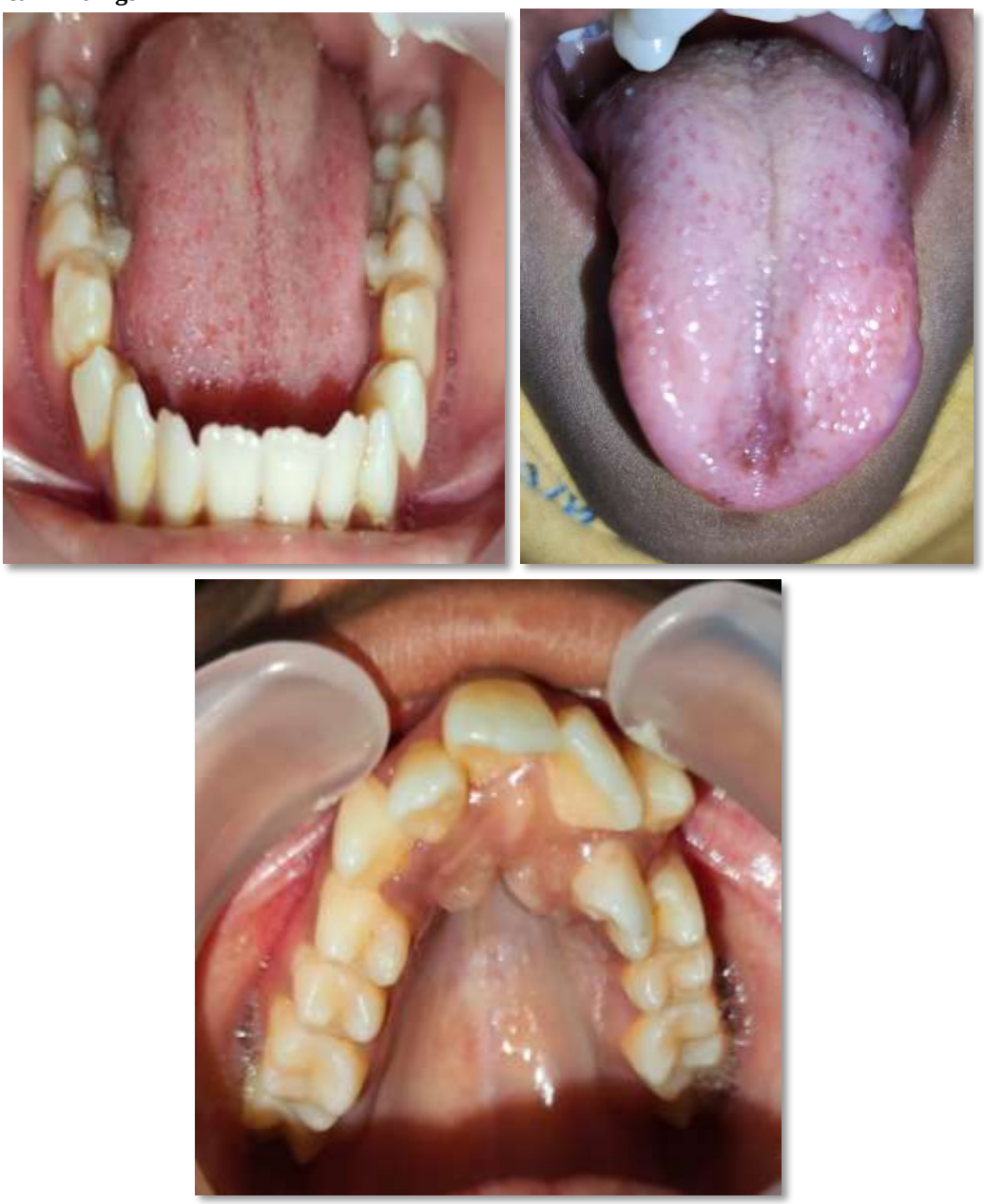


Fig 5:- Intraoral views showing crowded maxillary arch with deep pseudocleft in midline and normally aligned mandibular teeth.

Lateral cephalograph showing Midface hypoplasia Maxillary Retrusion or Hypoplastic Maxilla

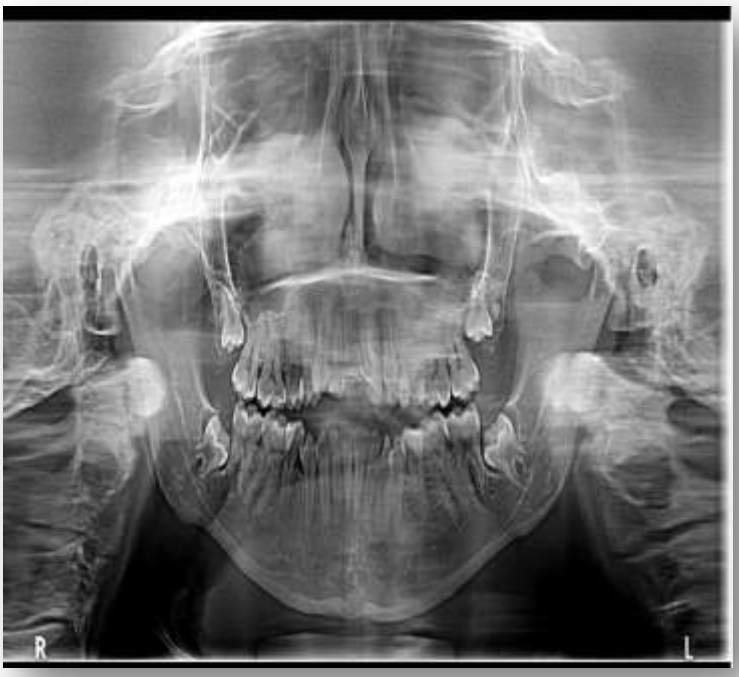
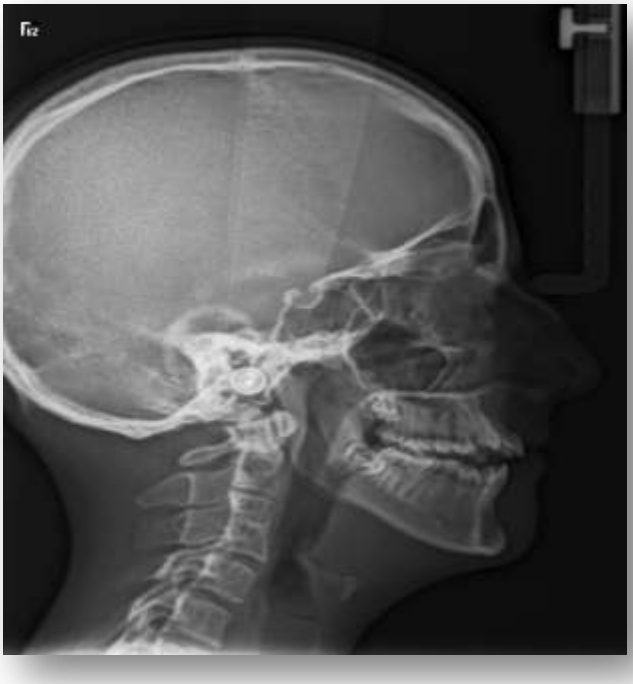


Fig 6:-Median pit on dorsalsurface of tongue.

A Panoramic radiograph showing Crowding, over retained and impacted teeth



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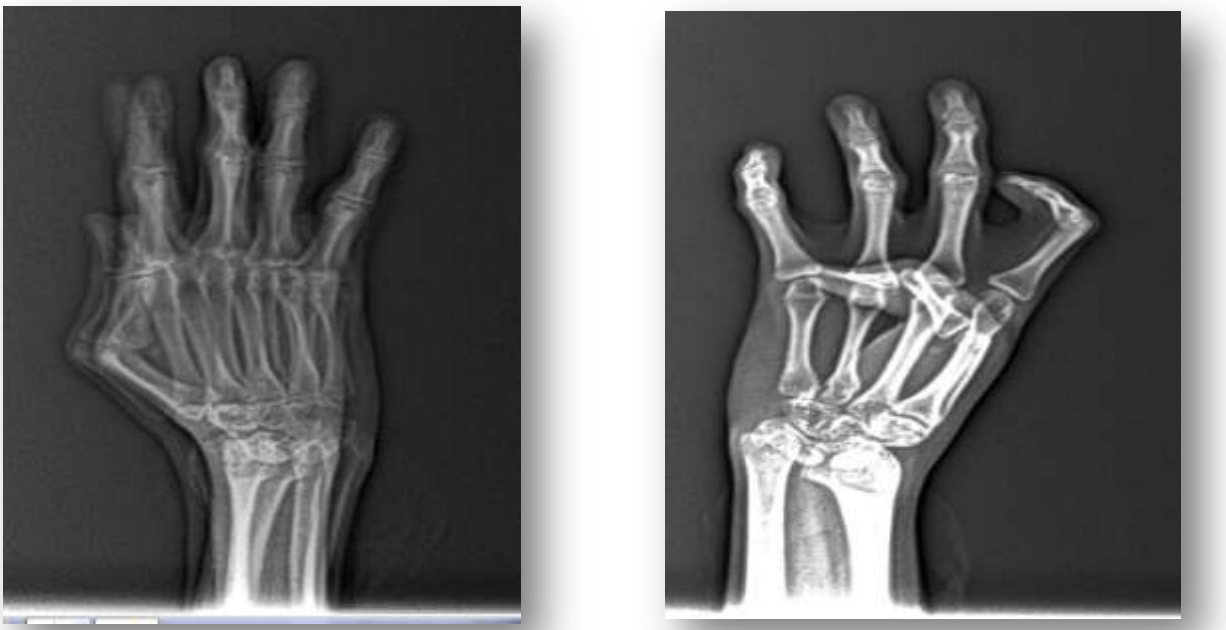

Hand wrist Radiograph showing soft tissue Syndactyly of hands with invandly hypoplastic thumb

Discussion:-

Genetic diseases have been on the rise, and this generally need to know for dentists in terms of properly diagnosing and dealing with these patients[3].Clinical features, diagnosis, and treatment of Apert syndrome, which is an autosomal dominant disorder caused by a mutation in the fibroblast growth factor receptor-2 (FGFR-2) gene [1,2,3,5,6]. Apert syndrome is characterized by craniosynostosis, which is the premature fusion of sutures in the skull, leading to abnormal head shape and facial features. Syndactyly, or fused digits, of the hands and feet is a distinguishing feature of Apert syndrome compared to other similar syndromes like Crouzon's, Carpenter, Pfeiffer, and Beare-Stevenson syndromes[1,2,3,7].Diagnosing Apert syndrome can be done prenatally using molecular genetic tests or confirmed at birth or during early infancy through clinical evaluation and specialized tests. Early diagnosis is crucial for initiating appropriate treatment, which requires a multidisciplinary approach involving various medical specialties such as respiratory, cerebral, dental, ophthalmic, and orthopedic care. Treatment options include surgical interventions to address craniosynostosis, syndactyly separation, and facial reconstruction for functional and aesthetic improvements[7,8].Apert syndrome patients often experience mental retardation, primarily due to increased intracranial pressure. Therefore, early craniotomy is necessary to prevent complications and relieve pressure. Orthodontic treatment and orthognathic surgery can correct malocclusion, and dental care is essential due to difficulties in maintaining oral hygiene[8]. Regular dental examinations, oral hygiene prophylaxis, fluoride treatments, and dental sealants can help prevent dental diseases.Prenatal diagnosis of Apert syndrome can be performed using common mutation sequencing on uncultured amniocytes, which is easier and less expensive than non-invasive prenatal diagnosis using high-throughput next-generation DNA sequencing. However, non-invasive prenatal diagnosis using foetal DNA from maternal plasma through polymerase chain reaction-based techniques has also been performed. It's important to note that the passage you provided seems to be a compilation of information from various sources and may not reflect the most up-to-date research or medical guidelines. It's always recommended to consult healthcare professionals or refer to current medical literature for accurate and reliable information[5,6].

Conclusion:-

Apert syndrome is a rare genetic disorder that affects craniofacial development, leading to craniosynostosis and distinctive facial features. Due to its rarity and unique characteristics, genotyping and genetic counselling are important for each diagnosed case. Genetic counselling helps individuals and families understand the inheritance

pattern and potential risks for future generations[1,2,4].Early diagnosis of craniosynostosis and timely surgical intervention are essential for favourable outcomes in individuals with Apert syndrome. Medical advancements have improved the life expectancy of patients with this condition. Multidisciplinary intervention, involving various medical specialties, is necessary to address the complex needs of these patients and improve their quality of life[9,10].Dentists also play a crucial role in the management of Apert syndrome. By focusing on parent and patient education, preventive care, and early detection and treatment of dental diseases, dentists can contribute to enhancing the overall well-being of individuals with this syndrome. Regular dental examinations, education on oral hygiene practices, and appropriate interventions can help improve oral health and promote better lives for these patients.It's important to note that the passage you provided appears to be a continuation of the previous discussion, highlighting the importance of genetic counselling, early diagnosis, and multidisciplinary care for individuals with Apert syndrome. However, it's always advisable to consult with healthcare professionals for accurate and personalized information regarding any medical condition[3].

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